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Alagille syndrome(ALGS) is an autosomal-dominant multisystem disorder affecting the liver, heart, face, eyes, skeleton and other organs. It is caused by mutations in one of two genes in the Notch signaling pathway, the Jagged1 (*JAG1*) gene or, in rare circumstances, the Notch homolog 2 (*NOTCH2*) gene. The object of this study is to evaluate combined genetic analyses with targeted next-generation sequencing (NGS), multiplex ligation probe amplification (MLPA) of *JAG1* and microarray comparative genomic hybridization (CGH) in subjects with ALGS, incomplete clinical features of ALGS and biliary atresia (BA). We recruited subjects from April 2013 to December 2015. All subjects underwent a targeted NGS analysis, including *JAG1* and *NOTCH2*. If no mutations were detected in *JAG1* or *NOTCH2*, or if copy number variations were suggested by the NGS analysis, we performed an MLPA analysis of *JAG1*. We also performed a microarray CGH analysis with whole-exon deletion detected by the MLPA analysis. Thirty subjects with ALGS, nine with incomplete ALGS and 17 with BA were enrolled, and detected pathogenic mutations in *JAG1* or *NOTCH2* in 24/30 subjects with ALGS and in 4/9 subjects with incomplete ALGS. No pathogenic mutations were detected in subjects with BA. The frequency of *JAG*1 mutations was as follows: single nucleotide variants (51.9%), small insertion or deletion (29.6%) and gross deletion (18.5%). The distribution of types of mutations in *JAG1* in this study was similar to that reported in a previous study, except for gross deletions, including partial-exon deletions and whole-exon deletions. Combined genetic analyses achieved efficient diagnostic yields for subjects with ALGS and incomplete ALGS.